

## SEQUENCE LISTING

## (1) GENERAL INFORMATION:

## (i) APPLICANT:

- (A) NAME: BASF Aktiengesellschaft
- (B) STREET: Carl-Bosch-Strasse 38
- (C) CITY: Ludwigshafen
- (E) COUNTRY: Federal Republic of Germany
- (F) POSTAL CODE: D-67056
- (G) TELEPHONE: 0621/6048526
- (H) TELEFAX: 0621/6043123
- (I) TELEX: 1762175170

(ii) TITLE OF APPLICATION: Method for diagnosing disorders by analysis of genes

(iii) NUMBER OF SEQUENCES: 2

## (iv) COMPUTER READABLE FORM:

- (A) MEDIUM TYPE: Floppy disk
- (B) COMPUTER: IBM PC compatible
- (C) OPERATING SYSTEM: PC-DOS/MS-DOS
- (D) SOFTWARE: PatentIn Release #1.0, Version #1.25 (EPA)

## (2) INFORMATION FOR SEQ ID NO: 1:

## (i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 1517 base pairs
- (B) TYPE: Nucleic acid
- (C) STRANDEDNESS: Double
- (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: cDNA for mRNA

(iii) HYPOTHETICAL: NO

(iv) ANTISENSE: NO

(vi) ORIGINAL SOURCE:

- (A) ORGANISM: Homo sapiens

(ix) FEATURES:

- (A) NAME/KEY: CDS
- (B) LOCATION: 1..1024

(xi) SEQUENCE DESCRIPTION: SEQ ID NO: 1:

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| ATG GGG GAG ATG GAG CAA CTG CGT CAG GAA GCG GAG CAG CTC AAG AAG | 48  |
| Met Gly Glu Met Glu Gln Leu Arg Gln Glu Ala Glu Gln Leu Lys Lys |     |
| 1 5 10 15   |     |
| CAG ATT GCA GAT GCC AGG AAA GCC TGT GCT GAC GTT ACT CTG GCA GAG | 96  |
| Gln Ile Ala Asp Ala Arg Lys Ala Cys Ala Asp Val Thr Leu Ala Glu |     |
| 20 25 30  |     |
| CTG GTG TCT GGC CTA GAG GTG GTG GGA CGA GTC CAG ATG CGG ACG CGG | 144 |
| Leu Val Ser Gly Leu Glu Val Val Gly Arg Val Gln Met Arg Thr Arg |     |
| 35 40 45  |     |
| CGG ACG TTA AGG GGA CAC CTG GCC AAG ATT TAC GCC ATG CAC TGG GCC | 192 |
| Arg Thr Leu Arg Gly His Leu Ala Lys Ile Tyr Ala Met His Trp Ala |     |
| 50 55 60  |     |
| ACT GAT TCT AAG CTG CTG GTA AGT GCC TCG CAA GAT GGG AAG CTG ATC | 240 |
| Thr Asp Ser Lys Leu Leu Val Ser Ala Ser Gln Asp Gly Lys Leu Ile |     |
| 65 70 75 80   |     |
| GTG TGG GAC AGC TAC ACC ACC AAC AAG GTG CAC GCC ATC CCA CTG CGC | 288 |
| Val Trp Asp Ser Tyr Thr Thr Asn Lys Val His Ala Ile Pro Leu Arg |     |
| 85 90 95  |     |
| TCC TCC TGG GTC ATG ACC TGT GCC TAT GCC CCA TCA GGG AAC TTT GTG | 336 |
| Ser Ser Trp Val Met Thr Cys Ala Tyr Ala Pro Ser Gly Asn Phe Val |     |

|            |             |            |            |            |             |            |            |             |            |     |     |     |     |     |     |      |
|------------|-------------|------------|------------|------------|-------------|------------|------------|-------------|------------|-----|-----|-----|-----|-----|-----|------|
| GCA        | TGT         | GGG        | GGG        | CTG        | GAC         | AAC        | ATG        | TGT         | TCC        | ATT | TAC | AAC | CTC | AAA | TCC | 384  |
| Ala        | Cys         | 115        | Gly        | Leu        | Asp         | Asn        | Met        | Cys         | Ser        | Ile | Tyr | Asn | Leu | Lys | Ser |      |
| CGT        | GAG         | GGC        | AAT        | GTC        | AAG         | GTC        | AGC        | CGG         | GAG        | CTT | TCT | GCT | CAC | ACA | GGT | 432  |
| Arg        | Glu         | Gly        | Asn        | Val        | Lys         | Val        | Ser        | Arg         | Glu        | Leu | Ser | Ala | His | Thr | Gly |      |
| TAT        | CTC         | TCC        | TGC        | TGC        | CGC         | TTC        | CTG        | GAT         | GAC        | AAC | AAT | ATT | GTG | ACC | AGC | 480  |
| Tyr        | Leu         | Ser        | Cys        | Cys        | Arg         | Phe        | Leu        | Asp         | Asp        | Asn | Asn | Ile | Val | Thr | Ser |      |
| TCG        | GGG         | GAC        | ACC        | ACG        | TGT         | GCC        | TTG        | TGG         | GAC        | ATT | GAG | ACT | GGG | CAG | CAG | 528  |
| Ser        | Gly         | Asp        | Thr        | Thr        | Cys         | Ala        | Leu        | Trp         | Asp        | Ile | Glu | Thr | Gly | Gln | Gln |      |
| AAG        | ACT         | GTA        | TTT        | GTG        | GGA         | CAC        | ACG        | GGT         | GAC        | TGT | ATG | AGC | CTG | GCT | GTG | 576  |
| Lys        | Thr         | Val        | Phe        | Val        | Gly         | His        | Thr        | Gly         | Asp        | Cys | Met | Ser | Leu | Ala | Val |      |
| TCT        | CCT         | GAC        | TTC        | AAT        | CTC         | TTC        | ATT        | TCG         | GGG        | GCT | TGT | GAT | GCC | AGT | GCC | 624  |
| Ser        | Pro         | Asp        | Phe        | Asn        | Leu         | Phe        | Ile        | Ser         | Gly        | Ala | Cys | Asp | Ala | Ser | Ala |      |
| AAG        | CTC         | TGG        | GAT        | GTG        | CGA         | GAG        | GGG        | ACC         | TGC        | CGT | GAG | ACT | TTC | ACT | GGC | 672  |
| Lys        | Leu         | Trp        | Asp        | Val        | Arg         | Glu        | Gly        | Thr         | Cys        | Arg | Gln | Thr | Phe | Thr | Gly |      |
| CAC        | GAG         | TCG        | GAC        | ATC        | AAC         | GCC        | ATC        | TGT         | TTC        | TTC | GCC | AAT | GGA | GAG | GCC | 720  |
| His        | Glu         | Ser        | Asp        | Ile        | Asn         | Ala        | Ile        | Cys         | Phe        | Phe | Pro | Asn | Gly | Glu | Ala |      |
| ATC        | TGC         | ACG        | GGC        | TCG        | GAT         | GAC        | GCT        | TCC         | TGC        | CGC | TTC | TTT | GAC | CTG | CGG | 768  |
| Ile        | Cys         | Thr        | Gly        | Ser        | Asp         | Asp        | Ala        | Ser         | Cys        | Arg | Leu | Phe | Asp | Leu | Arg |      |
| GCA        | GAC         | CAG        | GAG        | CTG        | ATC         | TGC        | TTC        | TCC         | CAC        | GAG | AGC | ATC | ATC | TGC | GGC | 816  |
| Ala        | Asp         | Gln        | Glu        | Leu        | Ile         | Cys        | Phe        | Ser         | His        | Gln | Ser | Ile | Ile | Cys | Gly |      |
| ATC        | ACG         | TCT        | GTG        | GCC        | TTC         | TCC        | CTC        | AGT         | GGC        | CGC | CTA | CTA | TTC | GCT | GGC | 864  |
| Ile        | Thr         | Ser        | Val        | Ala        | Phe         | Ser        | Leu        | Ser         | Gly        | Arg | Leu | Leu | Phe | Ala | Gly |      |
| TAC        | GAC         | GAC        | TTC        | AAC        | TGC         | AAT        | GTC        | TGG         | GAC        | TCT | ATG | AAG | TCT | GAG | CGT | 912  |
| Tyr        | Asp         | Asp        | Phe        | Asn        | Cys         | Asn        | Val        | Trp         | Asp        | Ser | Met | Lys | Ser | Glu | Arg |      |
| GTG        | GGC         | ATC        | CTC        | TCT        | GGC         | CAC        | GAT        | AAC         | AGG        | GTC | AGC | TGC | CTG | GGA | GTC | 960  |
| Val        | Gly         | Ile        | Leu        | Ser        | Gly         | His        | Asp        | Asn         | Arg        | Val | Ser | Cys | Leu | Gly | Val |      |
| ACA        | GCT         | GAC        | GGG        | ATG        | GCT         | GTG        | GCC        | ACA         | GGT        | TCC | TGG | GAC | AGC | TTC | CTC | 1008 |
| Thr        | Ala         | Asp        | Gly        | Met        | Ala         | Val        | Ala        | Thr         | Gly        | Ser | Trp | Asp | Ser | Phe | Leu |      |
| AAA        | ATC         | TGG        | AAC        | TGA        | G           | GAGGCTGGAG | AAAGGGAAAT | GGAAAGGCAGT | GAACACACTC |     |     |     |     |     |     | 1064 |
| Lys        | Ile         | Trp        | Asn        | *          |             |            |            |             |            |     |     |     |     |     |     |      |
| AGCAGCCCCC | TGCCCCGACCC | CATCTCATTC | AGGTGTTCTC | TTCTATATTC | CGGGGTGCCAT |            |            |             |            |     |     |     |     |     |     | 1124 |
| TCCCACTAAG | CTTTCTCCTT  | TGAGGGCAGT | GGGGAGCATT | GGACTGTGCC | TTTGGGAGGC  |            |            |             |            |     |     |     |     |     |     | 1184 |
| AGCATCAGGG | ACACAGGGGC  | AAAGAACTGC | CCCATCTCTT | CCCATGGCCT | TCCCTCCCCA  |            |            |             |            |     |     |     |     |     |     | 1244 |
| CAGTCCTCAC | AGCCTCTCCC  | TTAATGAGCA | AGGACAACCT | CCCCCTCCCC | AGCCCTTTGC  |            |            |             |            |     |     |     |     |     |     | 1304 |
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## (2) INFORMATION FOR SEQ ID NO: 2:

## (i) SEQUENCE CHARACTERISTICS:

(A) LENGTH: 341 amino acids

(B) TYPE: Amino acid

(D) TOPOLOGY: linear

(ii) MOLECULE TYPE: Protein

(xi) SEQUENCE DESCRIPTION: SEQ ID NO: 2:

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      35           40           45
Arg Thr Leu Arg Gly His Leu Ala Lys Ile Tyr Ala Met His Trp Ala
      50           55           60
Thr Asp Ser Lys Leu Leu Val Ser Ala Ser Gln Asp Gly Lys Leu Ile
      65           70           75           80
Val Trp Asp Ser Tyr Thr Thr Asn Lys Val His Ala Ile Pro Leu Arg
      85           90           95
Ser Ser Trp Val Met Thr Cys Ala Tyr Ala Pro Ser Gly Asn Phe Val
      100          105          110
Ala Cys Gly Gly Leu Asp Asn Met Cys Ser Ile Tyr Asn Leu Lys Ser
      115          120          125
Arg Glu Gly Asn Val Lys Val Ser Arg Glu Leu Ser Ala His Thr Gly
      130          135          140
Tyr Leu Ser Cys Cys Arg Phe Leu Asp Asp Asn Asn Ile Val Thr Ser
      145          150          155          160
Ser Gly Asp Thr Thr Cys Ala Leu Trp Asp Ile Glu Thr Gly Gln Gln
      165          170          175
Lys Thr Val Phe Val Gly His Thr Gly Asp Cys Met Ser Leu Ala Val
      180          185          190
Ser Pro Asp Phe Asn Leu Phe Ile Ser Gly Ala Cys Asp Ala Ser Ala
      195          200          205
Lys Leu Trp Asp Val Arg Glu Gly Thr Cys Arg Gln Thr Phe Thr Gly
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      225          230          235          240
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      245          250          255
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      260          265          270
Ile Thr Ser Val Ala Phe Ser Leu Ser Gly Arg Leu Leu Phe Ala Gly
      275          280          285
Tyr Asp Asp Phe Asn Cys Asn Val Trp Asp Ser Met Lys Ser Glu Arg
      290          295          300
Val Gly Ile Leu Ser Gly His Asp Asn Arg Val Ser Cys Leu Gly Val
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# SEQUENCE LISTING

<110> Siffert, Winfried

<120> THE USE OF A GENETIC MODIFICATION IN THE GENE FOR HUMAN  
G PROTEIN b3 SUBUNIT FOR THE DIAGNOSIS OF DISEASES

<130> 1135-2

<140> 09/180,783

<141> 1999-03-17

<150> PCT/EP97/02250

<151> 1997-05-02

<150> DE 19619362.1

<151> 1996-05-14

<160> 4

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<213> Homo sapiens

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 35 40 45

Arg Thr Leu Arg Gly His Leu Ala Lys Ile Tyr Ala Met His Trp Ala  
 50 55 60

Thr Asp Ser Lys Leu Leu Val Ser Ala Ser Gln Asp Gly Lys Leu Ile  
 65 70 75 80

Val Trp Asp Ser Tyr Thr Thr Asn Lys Val His Ala Ile Pro Leu Arg  
 85 90 95

Ser Ser Trp Val Met Thr Cys Ala Tyr Ala Pro Ser Gly Asn Phe Val  
 100 105 110

Ala Cys Gly Gly Leu Asp Asn Met Cys Ser Ile Tyr Asn Leu Lys Ser  
 115 120 125

Arg Glu Gly Asn Val Lys Val Ser Arg Glu Leu Ser Ala His Thr Gly  
 130 135 140

Tyr Leu Ser Cys Cys Arg Phe Leu Asp Asp Asn Asn Ile Val Thr Ser  
 145 150 155 160

Ser Gly Asp Thr Thr Cys Ala Leu Trp Asp Ile Glu Thr Gly Gln Gln  
 165 170 175

Lys Thr Val Phe Val Gly His Thr Gly Asp Cys Met Ser Leu Ala Val